bio392-cnv.frq

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Tumor information

Breast Carcinom: Breast Carcinoma is the most common Cancer in Women. It can also occur in men but it's far more likely in woman. Cancer airises when breast cells start to grow abnormally. Aberration leading to activation of oncogens and loss of tumor-suppressors are thought to be early breast cancer driver. Cells divide more rapildy than healthy cells and do accumulate and form an oddly shaped tissue. Because it needs constantly more space, when its limited is exceeded it begins to spread (metastasize). Hormonal, lifestyle and environmental factors may increase your risk of breast cancer. The likelihood of breast cancer have been identified with an increase in specific chromosmal gen mutation.

The NCIt codes is C9245

Access the CNV frequency Data

load library

```
library(pgxRpi)
```

Access Data

```
freq <- pgxLoader( type='frequency',output = 'pgxfreq', filters = "NCIT:C9245")
```

Metadata information is

```
freq$meta
```

```
##         code                      label sample_count
## 1 NCIT:C9245 Invasive Breast Carcinoma         3796
## 2      total                                   3796
```

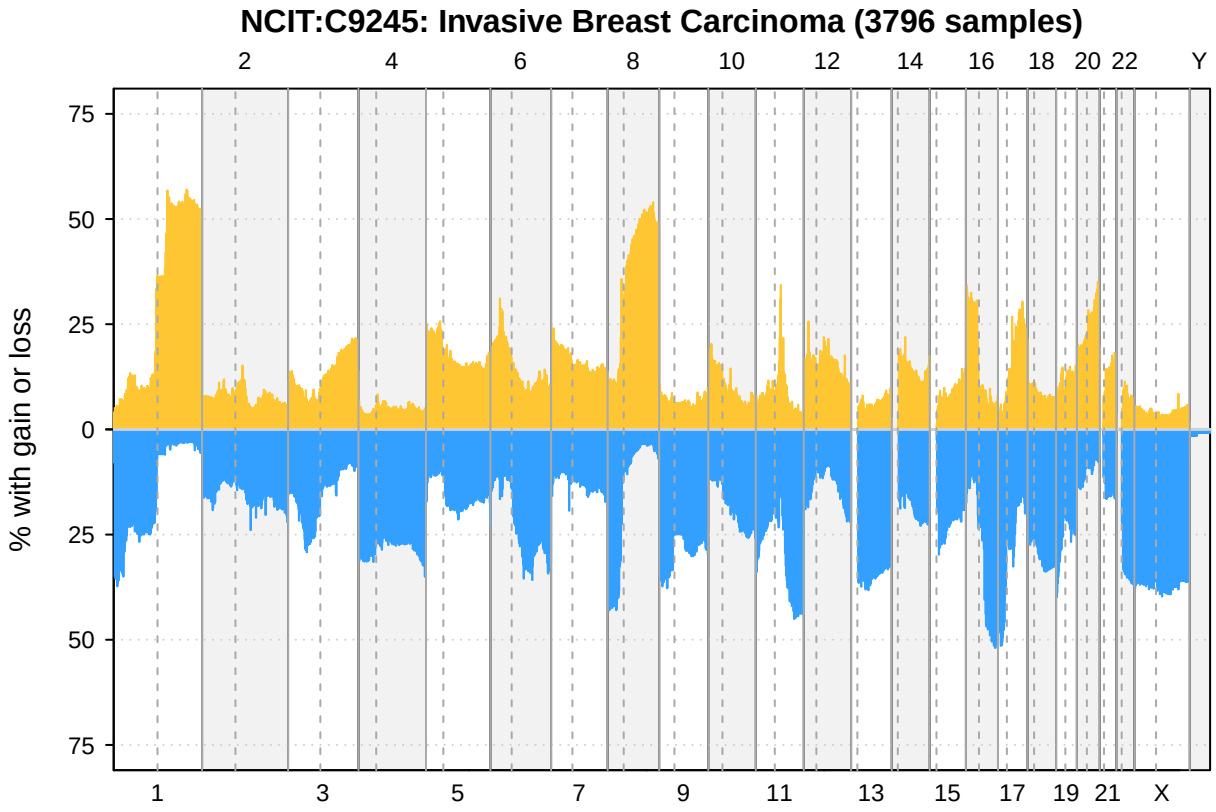
The data looks like this

```
freq$data$'NCIT:C9245'[c(1:3),]
```

```
##      filters reference_name   start     end gain_frequency loss_frequency no
## 1 NCIT:C9245              1       0  400000          2.397          1.159  1
## 2 NCIT:C9245              1  400000 1400000          4.083          7.877  2
## 3 NCIT:C9245              1 1400000 2400000          2.608          6.981  3
```

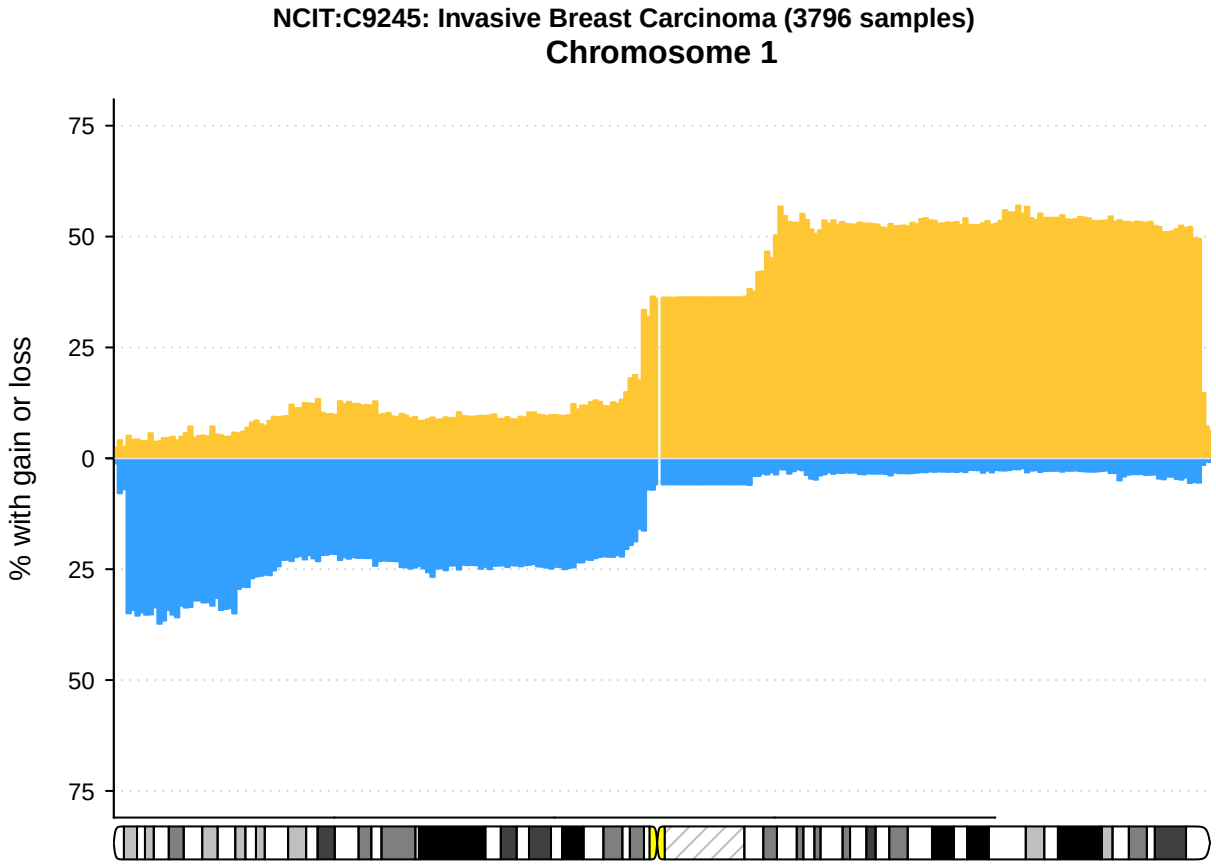
Visualisation

```
pgxFreqplot(freq)
```



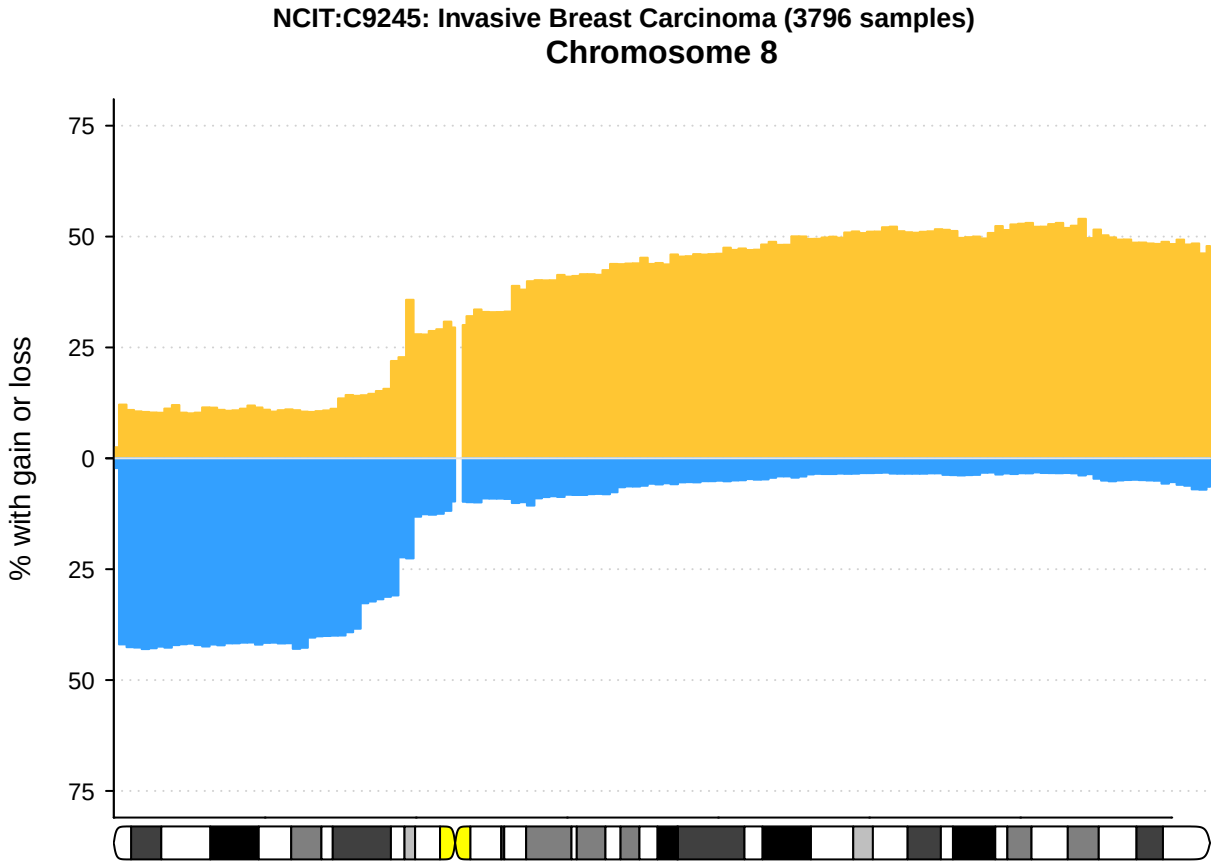
On the Upperside of the Graph the duplication of specific Chromosoms are shown in orange.
On the lowerside in blue the is illustrated the loss of DNA on the referred chromosom.

```
pgxFreqplot(freq, chrom=1)
```

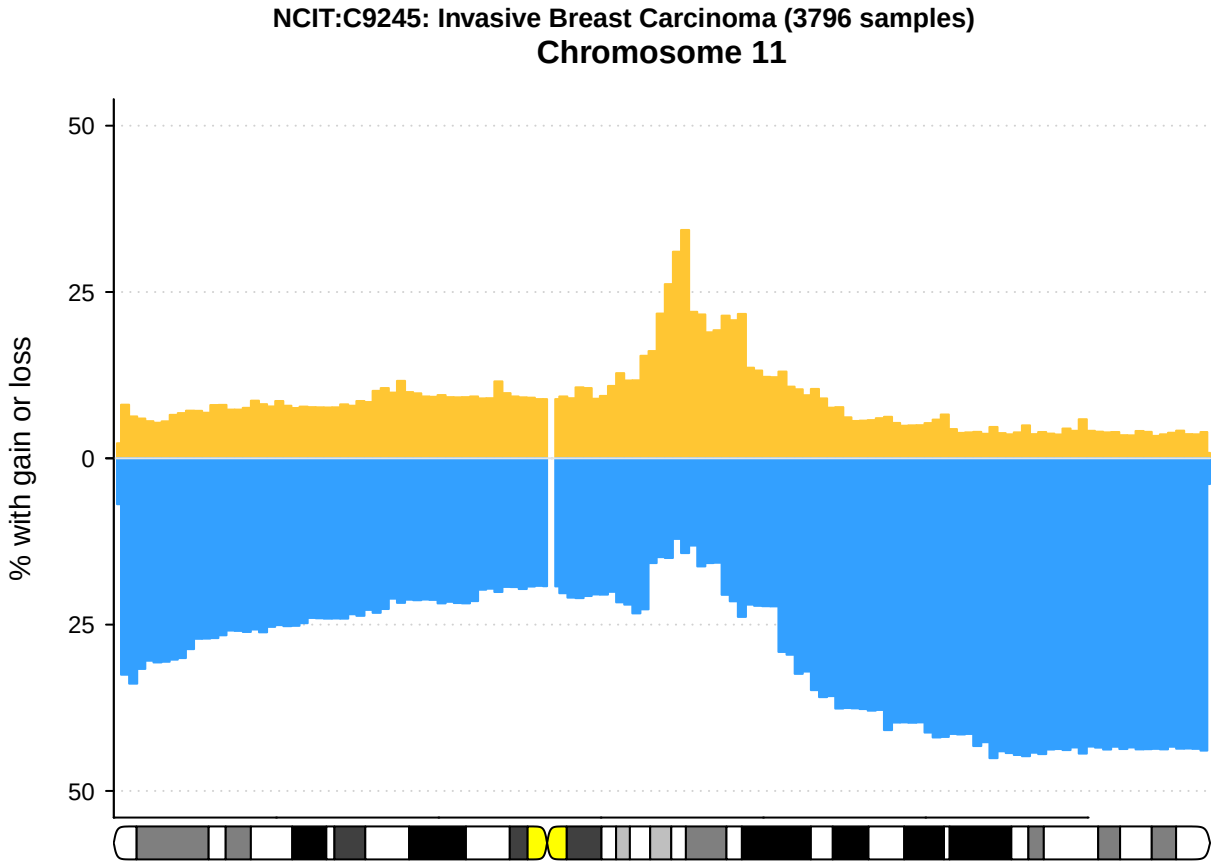


The impact on the Chromosom 1 seems to be important in case of breast Cancer. The Strongest alteration

```
pgxFreqplot(freq, chrom=8)
```




```
pgxFreqplot(freq, chrom=11)
```



Analysis

According to the plot, there are frequent gains for
on chromosom 1q, 8q, 16p, 20q amplification
and frequent deletion on chromosom: 1p, 4pq, 6q, 8p, 9pq, 11q, 13q, 15q, 22q, Xpq
with a treshold of 25% as cut-off.
On the Study:
[Genomic alterations during the in situ to invasive ductal breast carcinoma transition shaped by the immune system][https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8026652/]
we compare the Alteration between our plot from NCIT thesaurus and the data from the study.

From the plot:
1q, 8q, 16p, 20q amplification
1p, 4pq, 6q, 8p, 9pq, 11q, 13q, 15q, 22q, Xpq

Study:
1q, 8q, 17q, 20q amplification
8p, 11q, 16q, 17p loss

Overlapping arms between Study and our Plot:
1q, 8q, 20q in amplification
8p, 11q in loss.

As a result in both data sets the chromosom 1,8,11,20 are detrimental

in relation to breast carcinoma.